pennsylvania
Cancer Coalition

Pennsylvania Cancer Control Genetics/Genomics Healthcare Provider Toolkit

Revised March 2025

pennsylvania
Cancer Coalition

Dear Healthcare Provider:

We hope you find the multi-component Genetics/Genomics Toolkit useful. This toolkit is designed to provide healthcare professionals with the tools and resources needed to effectively integrate hereditary cancer risk assessment into practice. It offers practical information on the role of genetics in care, supports the recognition and appropriate referral of individuals at risk for hereditary cancer syndromes, and provides resources for discussing risk factors with patients.

The resources within the toolkit were assembled by members of the Genetics/Genomics Workgroup of the Pennsylvania Cancer Coalition (PCC), which works to integrate genetics into Pennsylvania's cancer control efforts. The Pennsylvania 2023-2033 Cancer Control Plan outlines a clear goal and comprehensive, evidence-based strategies for advancing access to genetic and genomic services. Incorporating genetics and genomics into cancer care is essential for early cancer diagnosis, effective treatment, and improved outcomes, ultimately reducing cancer incidence, morbidity, and mortality.

Much work remains to eliminate barriers and expand access to genetic and genomic services to create a healthier Pennsylvania and reduce the state's cancer burden. We invite you to join us in this important effort to make a meaningful impact. Please visit pacancercoalition.org for more information on the PCC Genetics/Genomics Workgroup and how to join the workgroup to support get involved this effort.

Andrea L. Durst, MS, DrPH, LCGC
PCC Genetics/Genomics Workgroup
Co-Chair Assistant Professor of Human Genetics
Licensed Genetic Counselor
University of Pittsburgh
adurst@pitt.edu

Rachel Schwiter, MGC, CGC
PCC Genetics/Genomics Workgroup Co-Chair
Assistant Program Director, Master of Science in Genetics Counseling
Licensed, Certified Genetic Counselor
Geisinger College of Health Sciences
rgschwiter1@geisinger.edu

pennsylvania
Cancer Coalition

Table of Contents

pennsylvania
Cancer Coalition

Acknowledgements:

Genetics/Genomics Workgroup Members

- **PCC Genetics/Genomics Workgroup Co-chairs**
  - **Durst, Andrea, MS, DrPH, LCGC**, Associate Director, Genetic Counseling Program and Co- Director, MPH in Public Health Genetics, University of Pittsburgh, Genetics/Genomics Workgroup Co-chair
  - **Rachel Schwiter, MGC, CGC,** Assistant Program Director, Master of Science in Genetic Counseling, Geisinger College of Health Sciences, Genetics/Genomics Workgroup Co-chair
  - **April Barry, LCSW, MSW, PN,** Evaluation Manger, Pennsylvania Department of Health, Comprehensive Cancer Control Program

- **PCC Genetics/Genomics Workgroup Members**

pennsylvania
Cancer Coalition

Pennsylvania Cancer Coalition
Healthcare Provider Genetics/Genomics Toolkit

Introduction

The Pennsylvania Cancer Coalition (PCC) is the statewide cancer coalition that engages stakeholders in implementing the 2023-2033 Pennsylvania Cancer Control Plan. Membership is open to all cancer stakeholders, including private and nonprofit organizations and individuals. In 2019, a Genetics/Genomics workgroup was formed within the Pennsylvania Cancer Coalition (PCC).

Based on the results of a statewide provider survey, the PCC Genetics/Genomics Workgroup identified the need for ongoing and updated provider education on genetic and genomic testing as well as the integration of the patient risk assessment into primary care, as its initial focus. Increasing healthcare providers' awareness of genetic testing requires access to appropriate education, including how to identify candidates for genetic counseling and testing and where to refer them when needed. The Genetics/Genomics Toolkit was created to enhance awareness of available resources for genetic counseling, testing, and services."

The 2023-2033 Pennsylvania Cancer Plan Objective 4:1 *Increase availability of and access to high quality cancer diagnosis and treatment* identifies populations of focus and strategies aimed at increasing availability and access to genetic counseling and testing. The strategies to achieve Objective 4:1 is multi-faceted and include community based and provider genetic hereditary syndrome education and advocating for insurance coverage, and the promotion of policy and systemic change to advance cancer genetics/genomics to save lives and improve health and quality of life.

This toolkit is designed to assist healthcare professionals with tools to collect a comprehensive family health history, helping them better understand an individual's health and risk factors. A family health history includes information about close relatives, including those who have passed away. Many diseases run in families, and shared environments or common exposures to risk factors can also contribute to health risks."

This toolkit is an interactive, curated, and annotated resource on hereditary cancer. It addresses genetic counseling and testing for hereditary cancer, advocacy and support organizations, risk assessment tools, current assessment and care guidelines, policy information, and general education for the public and healthcare

providers.

How to Collect a Family History

- **Collecting a family history | American Medical Association**

Risk Assessment and Screening Toolkit for Primary Care Providers

- **Risk Assessment and Screening Toolkit to Detect Familial, Hereditary and Early Onset Colorectal Cancer - American Cancer Society National Colorectal Cancer Roundtable**

This step-by-step toolkit aims to improve the ability of primary care clinicians to systematically collect, document, and act on a family history of CRC and adenomas polyp

Elements of Informed Consent

- **Informed Consent and Pretest Counseling:**
  - See example from: Pretest Counseling Key Points (jax.org)

Family Health History Tools for Patients

Patients can input family history details, and the following tools will create family history.

- **My Family Health Portrait: A Tool from the Surgeon General**

https://phgkb.cdc.gov/FHH/html/index.html

My Family Health Portrait is a family history drawing tool developed by the Surgeon General's Office and maintained by the CDC. Individuals can enter their family history, see how conditions run in their family, and use the risk assessment tools. In addition, the family history can be saved, printed, and shared with a healthcare provider.

- **Susan G. Komen Family Health History**

https://www.komen.org/fhht/index.html

This tool allows people to enter in their family history information for each blood relative. The tool contains a link to more information about risk factors for breast cancer, allows the user to save their family history, and allows the user to share the family history with another relative. No family tree is drawn with this tool.

- **It Runs in My Family**

https://itrunsinmyfamily.com/

This family history tool utilizes a chatbot that walks the user through a set of questions to gather their family history. An individual must get through the entire set of questions for a report to be emailed. The questions are straightforward and understandable.

Resources providers can recommend to patients on how to gather family health history

information:

- **Family History - National Society of Genetic Counselors**
  https://www.aboutgeneticcounselors.org/Resources-to-Help-You/Post/how-to-draw-your- family-tree

This resource from the National Society of Genetic Counselors offers tips on how to draw out a family tree (pedigree). It includes a list of conditions and details that are particularly important to note when writing out a family tree and other relevant details health care providers may ask about family history. This webpage links to https://www.aboutgeneticcounselors.org/Resources-to-Help-You/Post/frequently-asked-questions-resources for additional details.

- **A guide to family health history – Geisinger Health**

https://www.geisinger.org/-/media/OneGeisinger/pdfs/ghs/research/family-
This pamphlet from Geisinger was compiled as a "manual" for collecting family health history information. Sample questions are listed for use as a starting point for conversations with family members.

Cancer Risk Assessment Tools

- **NCI Gail model**
  https://bcrisktool.cancer.gov/index.html
  The Gail model considers a woman's personal medical history, familial history, and reproductive history. Specifically, it includes current age, age at menarche, age at first live birth, number of biopsies (0, 1, 2 or more), biopsy outcome (atypical hyperplasia: yes, no, unknown), and number of first-degree relatives (mothers/sisters/daughters) with breast cancer (none, one, more than one, unknown).
  Limitations to the Gail model:
  should not be used in women with:
  - a history of breast cancer, DCIS or LCIS,
  - a known hereditary predisposition to breast cancer,
  - a significant family history suggestive of a hereditary predisposition.

  This model considers only first-degree relatives and does not include the paternal history of breast cancer or male breast cancer, history of ovarian cancer, and age at cancer diagnosis.

  The Gail Model, first published in 1989, was developed to predict the risk of invasive breast cancer and carcinoma in situ. It was later refined for more precise risk assessment and validated in 1999 using data from the Breast Cancer Prevention Trial. Updates adjusted risk estimates for black, Asian, Pacific Islander, and Hispanic women. However, it may underestimate risk for black women with prior biopsies and Hispanic women born outside the

  U.S. Due to limited data, risk estimates for American Indian/Alaska Native women are based

partly on white women's data and may be less accurate. Mathematically, it is an unconditional logistic regression model that provides a ratio of risk in women with specified risk factors compared to women's risk without risk factors. The base race-specific population

risk is derived from SEER estimates.

- **IBIS**
  http://www.ems-trials.org/riskevaluator/
  The International Breast Cancer Intervention Study (IBIS) tool is a risk-calculating model used to estimate a person's likelihood of carrying a pathogenic variant in BRCA1 or BRCA2. It also estimates the likelihood of developing breast cancer in the following ten years and over the individual's lifetime (up to age 80).

  The model was developed with data from the International Breast Cancer Intervention Study (IBIS), including a cohort of daughters of patients diagnosed with breast cancer. The input for the model development was the estimated probability of carrying a BRCA1 or BRCA2 pathogenic variant and the estimated breast cancer risks based on family history, hormonal and reproductive factors, and personal characteristics.

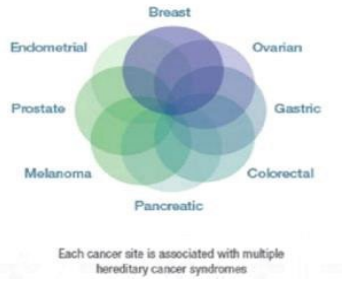
  The risks of developing breast cancer for the general population were taken from data on the first breast cancer diagnosis (ICD-10 code C50) in the Thames Cancer Registry area (UK) between 2005-2009. The risk from family history (caused by the adverse genes) is modeled based on the findings from a Swedish population-based study on cancer risk associated with a family history of breast and/or ovarian cancer in mothers. As the model incorporates the BRCA genes and a low penetrance gene, family history is used in conjunction with Bayes' theorem to iteratively produce the likelihood of carrying any genes predisposing to breast cancer, which in turn affects the chance of developing breast cancer. This risk was further refined based on the woman's personal history, including age, age at menarche, age at first-child, body mass index, benign breast disease (hyperplasia, atypical hyperplasia, LCIS), age at menopause, and use of hormone replacement therapy. The latest version of the model (v8) incorporates mammographic density (applicable only for those age 40 and older).

  ***Note of caution*:** The IBIS model tends to overestimate breast cancer risk in the presence of atypia (Boughey et al.) though the model was subsequently revised to correct for this. Also, the risk associated with mammographic density is included in the estimates for those older than 40; a study shows that mammographic density is not associated with subsequent breast cancer risk for those with atypical hyperplasia. Thus, the model might not be appropriate to use for women with atypia.

- **CanRisk**
  https://canrisk.org/

BOADICEA (CanRisk) is a comprehensive model that can be used to calculate the future risks of developing breast or ovarian cancer using information obtained from family history, lifestyle/hormonal risk factors, rare pathogenic variants in moderate, high-risk breast/ovarian cancer susceptibility genes, common breast/ovarian cancer genetic susceptibility variants (Polygenic Risk Scores), and mammographic density. It can also calculate the likelihood of carrying mutations in the moderate to high-risk genes BRCA1, BRCA2, PALB2, ATM, and CHEK2.

- Registration to create an account is required to access the tool.



Hereditary Cancer Syndromes – General

- **NYMAC Cancer Genetics Referrals: When to Refer to Genetics**
  https://www.wadsworth.org/cancer-referrals
  This resource provides a brief review of red flags that would indicate a patient may need a referral to genetic cancer services.

- **Genetic and Rare Disease Information Center (GARD) Hereditary Cancer Syndromes**
  https://rarediseases.info.nih.gov/diseases/diseases-
  This website is a landing page that links to more in- depth descriptions of various hereditary cancer syndrome summaries included on the GARD website. Information on symptoms, inheritance, cause, diagnosis and treatment for each condition are reviewed.

- **GeneReviews**
  http://www.genereviews.org
  GeneReviews provides detailed reviews of multiple cancer syndromes, which can be searched from the link to the homepage.
  The *GeneReviews* Website offers an international point-of-care resource for busy clinicians with clinically relevant and medically actionable information on inherited conditions in a standardized journal-style format. Information is provided on diagnosis, management, and genetic counseling for patients and their families.
  Each chapter in *GeneReviews* is written by one or more experts on the specific condition or disease and goes through rigorous editing and peer review process before being published online.

- **National Society of Genetic Counselors (NSGC): About Genetic Counselors**
  https://www.aboutgeneticcounselors.org/Resources-to-Help-You/Post/hereditary-cancer-hereditary-breast-and-ovarian-cancer-and-brca1-and-brca2
  This site contains general information, videos, and other linked resources on hereditary cancer syndromes. The website is patient-friendly and may be helpful to providers as they talk to patients about genetic counseling and testing for hereditary cancer syndromes.

Lynch Syndrome

- **GeneReviews:** Lynch syndrome https://www.ncbi.nlm.nih.gov/books/NBK1211/ GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition**.**
- **Lynch Syndrome Screening Network (LSSN)** https://www.lynchscreening.net/
  The Lynch Syndrome Screening Network promotes universal tumor screening for Lynch syndrome. The website contains helpful information about Lynch syndrome, screening for Lynch syndrome, and tools for implementing universal tumor screening.
- **Collaborative Group of the Americas on Inherited Gastrointestinal Cancers (CGA-IGC)** https://www.cgaigc.com/
  The CGA-IGC's mission is to advance the science and clinical care of inherited gastrointestinal cancers through research and education as the leading authority in the Americas. Their website contains a registry tool, a clinic tool, and a resource library.

Hereditary Breast and Ovarian Cancer Syndrome

- **GeneReviews: BRCA1 and BRCA2-Associated Hereditary Breast and Ovarian Cancer** https://www.ncbi.nlm.nih.gov/books/NBK1247/
  GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition.
- **GeneReviews: PTEN Hamartoma Tumor Syndrome (Cowden syndrome)** https://www.ncbi.nlm.nih.gov/books/NBK1488/
  GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition.
- **GeneReviews: LiFraumeni syndrome** https://www.ncbi.nlm.nih.gov/books/NBK1311/
  GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition.
- **BRCA Exchange** https://brcaexchange.org/
  The BRCA Exchange site provides information about the pathogenicity of *BRCA1* and *BRCA2* variants that have been curated by ENIGMA, an international expert panel.

pennsylvania
Cancer Coalition

Guidelines

- **NCCN Guidelines**
  https://www.nccn.org/professionals/physician_gls/default.aspx **The** National Comprehensive Cancer Network provides clinical practice guidelines related to multiple hereditary cancer syndromes, including hereditary breast and ovarian cancer syndromes and hereditary colorectal cancer syndromes. To access the guidelines, you must sign up for a free account.

- **USPSTF Recommendations: BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing**
  https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing
  Updated in 2019, guidance on when to utilize familial risk assessment tools for women who have a personal or family history of breast, ovarian, and/or primary peritoneal cancer. These guidelines do not consider risk assessment for men with a personal or family history of breast cancer.

- **American College of Medical Genetics and Genomics and the National Society of Genetic Counselors: Referral Indications for Cancer Predisposition Assessment**
  https://www.acmg.net/docs/ACMG_Practice_Guideline_Referral_Indications_for_cancer_predisposition.pdf
  These practice guidelines provide information on when to refer to genetic counseling for several different hereditary cancer syndromes. The guidelines are organized by cancer type and offer brief explanations for many hereditary cancer syndromes.

- **ACG Clinical Guideline: Genetic Testing and Management of Hereditary Gastrointestinal Cancer Syndromes**
  https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4695986/
  These Guidelines from the American College of Gastroenterology include genetic testing and management of Lynch syndrome, FAP, attenuated FAP (AFAP), MUTYH-associated polyposis (MAP), Peutz-Jeghers syndrome, juvenile polyposis syndrome, Cowden syndrome, serrated (hyperplastic) polyposis syndrome, hereditary pancreatic cancer, and hereditary gastric cancer.

Find a Genetic Provider Links

- **National Society of Genetic Counselors (NSGC) Find a Genetic Counselor**
  https://findageneticcounselor.nsgc.org
  This directory can be used to find a genetic counselor by city, state, or zip code and specialty area.

- **American College of Medical Genetics and Genomics (ACMG): Find a Genetic Clinic**
  https://clinics.acmg.net/

pennsylvania
Cancer Coalition

This directory contains a listing of genetics clinics, including those that offer telegenetics. The directory is searchable by city, state, and specialty area. It is a comprehensive directory that is not linked to organization membership and is updated regularly.

- **OrphaNet**

https://www.orpha.net/consor/cgi-bin/Clinics.php?lng=EN
OrphaNet is a portal for rare diseases and orphan drugs. Their directory allows the user to search for an expert center by condition and country.

- **American Board of Genetic Counseling (ABGC): Find a Genetic Counselor**

https://abgc.learningbuilder.com/Search/Public/MemberRole/Verification
A searchable directory of all board-certified genetic counselors

- **Pennsylvania Association of Genetic Counselors (PAGC)**

https://www.pennsylvaniagc.org/patients-and-providers
Listing of genetics clinics in PA. Email for questions: pennsylvaniagc@gmail.com

Genetic Discrimination, Cost, Insurance Coverage

- **US Equal Employment Opportunity Commission Information on the Genetic Information and Non-Discrimination Act of 2008 (GINA)**
  https://www.eeoc.gov/laws/statutes/gina.cfm
- This website contains the Genetic Information and Non-Discrimination Act of 2008.

- **GINA Fact Sheet for Patients and Providers**
  https://www.eeoc.gov/laws/guidance/fact-sheet-genetic-information- nondiscrimination-act

- Additional resources on GINA https://ginahelp.org/

PA Act 1 2023

Act 1 of 2023 - PA Breast Cancer Coalition
This Act eliminates all costs associated with screening breast MRI, ultrasound, genetic testing and counseling for individuals at high risk and insured under Pennsylvania law (state-regulated insurance). https://www.pabreastcancer.org/act-1-of-2023/

Direct-to-Consumer Testing

- Patients can purchase a test kit directly from a company without a healthcare professional's involvement

pennsylvania
Cancer Coalition

- **GeneReviews: Resources for Genetics Professionals – Direct-to-Consumer Genetic Testing** https://www.ncbi.nlm.nih.gov/books/NBK542335/
  GeneReviews provides detailed reviews written by experts in the field.

- **National Human Genome Research Institute: Direct-to-Consumer Genetic Testing**
  https://www.genome.gov/dna-day/15-ways/direct-to-consumer- genomic testing
  This website provides general information about direct-to-consumer genetic testing and links to multiple other resources for patients and providers on the topic.

- **Watershed DNA https://www.watersheddna.com/**
  Founded by a genetic counselor, this website provides several resources for individuals who have undergone direct-to-consumer genetic testing, including tests for hereditary cancer. In addition, the website includes options for a consultation with a genetic counselor, a blog, an online support group, and additional resources.

- **My Gene Counsel https://www.mygenecounsel.com/**
  Founded by a genetic counselor, this company offers services to provide genetic counseling information and ongoing updates for patients and providers.

CME Opportunities in Cancer Genetics

- **City of Hope Course** https://www.cityofhope.org/education/health- professional-education/cancer-genomics-education- program/intensive-course-in-cancer-risk-assessment- overview

- **International Symposium on Hereditary Breast and Ovarian Cancer**
  https://www.brcasymposium.ca/

- **Cancer genetic certification program from National Consortium of Breast Centers**
  https://www.cgracertification.org/certification/

- **Basser Center Scientific Symposium**
  https://www.basser.org/investigators-clinicians/scientific-symposium

- **American Society of Clinical Oncology (ASCO) Genetics Toolkit**
  https://www.asco.org/practice-policy/cancer-care-initiatives/genetics-toolkit

- **International Society of Nurses in Genetics (ISONG) Webinars**
  https://www.isong.org/page-1325047

- **Collaborative Group of the Americas Inherited Gastrointestinal Cancer Annual Conference and Lectureships**
  https://www.cgaigc.com/events

- **National Coordinating Center**

pennsylvania
Cancer Coalition

Evidence-Based Guidelines 101 - National Coordinating Center for the Regional Genetics Networks

Somatic Tumor Testing Resources

Somatic mutation profiling aims to identify driver mutations that can be targeted for treatment. However, it is not a substitute for germline testing, as its sensitivity is generally lower than dedicated germline tests. Additionally, a negative result in somatic testing does not rule out the presence of a germline mutation. Germline variants may be incidentally found in somatic testing. In this case, patients should be referred for genetic counseling and confirmatory germline testing. A patient who meets the criteria for germline testing should be referred for genetic counseling and confirmatory germline testing whether the patient has had somatic mutation profiling or not.Below is a list of websites that can be used to provide helpful information about individual mutations.

Somatic only

- **My Cancer Genome**
  https://mycancergenome.org/
  My Cancer Genome contains information on the clinical impact of cancer-related genes,

  proteins, and other biomarker types on the use of anticancer therapies in cancer. There are associated genetic biomarkers, diseases, and pathways. Links to applicable clinical trials are available.

- **Clinical Interpretation of Variants in Cancer (CIViC)**
  https://civicdb.org/
  CIViC is an open access, open-source, community-driven web resource disseminating knowledge about clinically significant cancer genome alterations. There are clinical interpretations of variants. Links to ClinVar and COSMIC are available.

- **Catalogue Of Somatic Mutations in Cancer (COSMIC)**
  https://cancer.sanger.ac.uk/cosmic/
  COSMIC is the world's largest and most comprehensive resource for exploring the impact of somatic mutations in human cancer. COSMIC can be searched by gene, cancer type, or mutation. Links to OMIM and NCBI EntrezGene are available.

Somatic and germline

- **ClinGen**
  https://clinicalgenome.org/
  ClinGen is an authoritative central resource that defines the clinical relevance of genes and variants for use in precision medicine and research. Genomic and phenotypic data is shared between clinicians, researchers, and patients through centralized and federated databases for clinical and research use. ClinGen has variant curation with gene-disease validity and clinical actionability. Links to ClinVar and Gene Review are

present.

- **ClinVar** https://www.ncbi.nlm.nih.gov/clinvar/
  ClinVar aggregates information about genomic variation and its relationship to human health. ClinVar is a public archive of reports of the relationships among human variations and phenotypes, with supporting evidence. ClinVar facilitates access to and communication about the relationships asserted between human variation and observed health status and the history of that interpretation. ClinVar processes submissions reporting variants found in patient samples, assertions about their clinical significance, information about the submitter, and other supporting data.

- **Online Mendelian Inheritance in Man (OMIM)**
  https://www.omim.org/
  OMIM is a comprehensive, authoritative catalog of human genes and genetic phenotypes that is freely available and updated daily by the Johns Hopkins University McKusick Nathans Department of Genetic Medicine. OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. The full-text referenced overviews in OMIM contain information on all known mendelian disorders and traits, and over 15,000 genes, focusing on the gene-phenotype relationships.

- **MedGen**
  https://www.ncbi.nlm.nih.gov/medgen
  MedGen is NCBI's portal to information about human disorders and other phenotypes having a genetic component. MedGen is structured to serve health care professionals, the medical genetics community, and other interested parties by providing centralized access to diverse types of content.

Pharmacogenomic Testing Applicable for Cancer Treatment

- **Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines**
  https://cpicpgx.org/guidelines/ https://www.pharmgkb.org/guidelineAnnotations
  CPIC addresses barriers to clinical implementation of pharmacogenetic tests by creating, curating, and posting freely available, peer-reviewed, evidence-based, updatable, and detailed gene/drug clinical practice guidelines. CPIC guidelines follow standardized formats, include systematic grading of evidence and clinical recommendations, use standardized terminology, are peer-reviewed, and published in a leading journal (in partnership with Clinical Pharmacology and Therapeutics) with simultaneous posting to cpicpgx.org, where they are regularly updated.

Patient Advocacy Organizations

- **Alive & Kickin'**
  https://www.aliveandkickn.org/
  Alive & Kickin' serves to improve the lives of individuals and families affected by Lynch

syndrome and associated cancers through research, education, and screening.

- **Association for Multiple Endocrine Neoplasia (MEN)Disorders**
  https://www.amend.org.uk/
  The Association for Multiple Endocrine Neoplasia Disorders (AMEND) serves to improve the well-being of all persons affected by MEN, and endocrine tumors. AMEND offers support to patients and families, education and supports MEN research.

- **American Multiple Endocrine Neoplasia Support**
  http://www.amensupport.org
  The American Multiple Endocrine Neoplasia Support is a voluntary organization whose mission is to provide education and support to patients, families, and medical personnel regarding multiple endocrine neoplasia (MEN) type 1, MEN type 2a, and MEN type 2b. Multiple Endocrine Neoplasia (MEN) type 1 is a rare genetic disorder were benign tumors form in various endocrine glands. MEN type 2 is another rare genetic condition that increases the risk of medullary thyroid carcinoma (a type of thyroid cancer) and causes benign tumors in other endocrine glands. Both types of MEN disrupt the normal function of endocrine glands, leading to hormonal imbalances that can cause conditions like diabetes and other health issues

- **Bright Pink**
  https://brightpink.org/
  Bright Pink helps save lives from breast and ovarian cancer by empowering women to know their risks and proactively manage their health.

- **Fanconi Anemia Research Fund, Inc.**
  http://www.fanconi.org/
  The Fanconi Anemia Research Fund is a nonprofit organization that raises funds for medical research into Fanconi anemia, an inherited condition that leads to a deficiency of certain blood cells that are produced by the bone marrow

- **FORCE**
  www.facingourrisk.org
  FORCE improves the lives of individuals and families affected by hereditary breast, ovarian, and related cancers. FORCE accomplishes this mission by creating awareness, supplying information and support to our community, advocating for, supporting research, and working with the research and medical communities to help people dealing with hereditary breast, ovarian, and related cancers.

- **Gorlin Syndrome Alliance**
  https://gorlinsyndrome.org/
  The mission of the Gorlin Syndrome Alliance is to support thoughtfully, comprehensively educate and aggressively seek treatments and a cure for Gorlin syndrome, its

manifestations, and sporadic BCCs.

- **Li-Fraumeni Syndrome Association**
  http://www.lfsassociation.org/
  LFS Association provides a wide range of information, advocacy, and support services for individuals and families with Li-Fraumeni Syndrome

- **Lynch Syndrome International**
  https://lynchcancers.com/
  The primary mission of Lynch Syndrome International (LSI) is to provide support for individuals afflicted with Lynch syndrome, create public awareness of the syndrome, educate members of the general public and health care professionals, and support for Lynch syndrome research endeavors.

- **Neurofibromatosis Network**
  http://www.nfnetwork.org
  The Neurofibromatosis, Inc. is a national, voluntary, nonprofit organization dedicated to providing information, support, and advocacy to individuals and family members affected by neurofibromatosis type I (NF1) and type II (NF2). NF1 is a genetic disorder characterized by the development of multiple benign tumors on the covering of nerve fibers and the appearance of brown spots and freckles on the skin. NF2 is a rare genetic disorder characterized by the development of benign tumors on both auditory nerves and in other areas of the body.

- **Neurofibromatosis Northeast**
  http://nfnortheast.org/
  The Mission of NF Northeast is to find a treatment and the cure for neurofibromatosis by promoting scientific research, creating awareness, and supporting those affected by NF.

- **PA Breast Cancer Coalition**
  https://www.pabreastcancer.org/
  The Pennsylvania Breast Cancer Coalition is a grassroots advocacy organization developed in 1993 with one mission in mind - “to help find a cure for breast cancer NOW… so our daughters don’t have to

- **No Stomach for Cancer**
  https://www.nostomachforcancer.org/
  No Stomach for Cancer is an organization that supports research and unites the caring power of people worldwide affected by stomach cancer. Their key priorities include 1) advancing awareness and education about stomach cancer, including Hereditary Diffuse Gastric Cancer (HDGC), 2) establishing and maintaining a network of support for affected families, 3) supporting research efforts for screening, early detection, treatment, and prevention of stomach cancer, and 4) sustaining a strong and high performing organization.

pennsylvania
Cancer Coalition

- **Pheo Para Alliance**
  http://www.pheopara.org
  Pheo Para Alliance invests in research to accelerate treatments and cures for pheochromocytoma and paraganglioma while empowering patients, their families, and

  medical professionals through advocacy, education, and a global community of support.

- **Sharsheret**
  https://sharsheret.org/brcagenetics/counseling-testing
  Sharsheret is a national nonprofit dedicated to supporting Jewish women and families at increased genetic risk for breast and ovarian cancer. They provide personalized support and life-saving educational outreach. While they specialize in young women and Jewish families, their programs are open to all women and men.

- **Tuberous Sclerosis Alliance**
  http://www.tsalliance.org
  The Tuberous Sclerosis Alliance (TSA) is a voluntary nonprofit organization dedicated to finding a cure for tuberous sclerosis complex (TSC) while improving the lives of those afflicted.

- **VHL Alliance**
  http://www.vhl.org
  The VHL Alliance is a nonprofit organization dedicated to improving diagnosis, treatment, and quality of life for individuals and families affected by von Hippel-Lindau disease (VHL). VHL is an inherited multisystem disorder characterized by the abnormal growth of blood vessels (angiomatosis) in certain body areas, such as the retinas, the brain, the spinal cord, and/or the adrenal glands.

General information about genetic counseling/testing (video links, etc.)

- **Genetic Counselors: Personalized Care for Your Genetic Health**
  https://www.aboutgeneticcounselors.org/
  This website contains basic information, including what a genetic counselor is, what to expect from a genetic counseling appointment, and answers to other basic patient questions. The website also contains several short videos that can be shared with patients.

Podcast Resources

- **Grey Genetics Patient Stories Podcast**
  https://www.greygenetics.com/podcast/
  This podcast shares the stories of patients and their experiences with genetic conditions and genetic counselors, including some episodes with individuals who have a hereditary cancer syndrome.